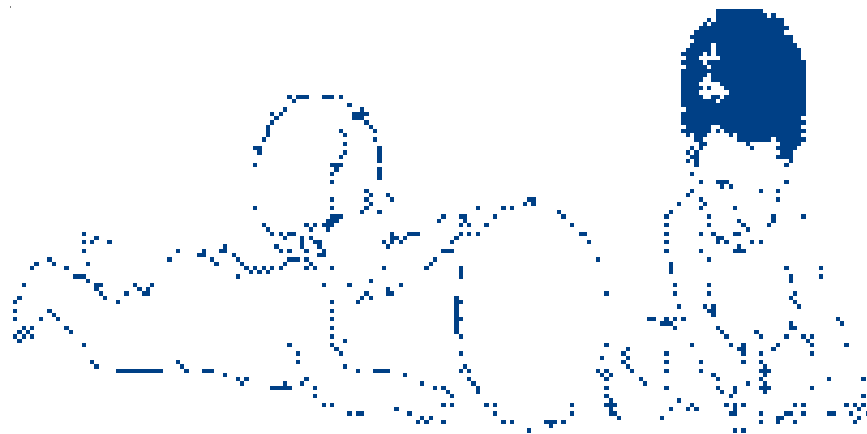




Nebraska's Plan for Newborn Screening and Genetics Services



This document can be found on the Internet at:

<http://www.hhs.state.ne.us/nsp/stateplan.pdf>

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This state plan was originally written in March 2003.

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Preface

Since the infancy of newborn screening programs in the 1960s and the more recent advances in the study of human genetics, the general public has become more aware of the availability of early detection and diagnosis of genetic disorders. With early detection and diagnosis of genetic conditions, treatments and therapies can begin sooner to potentially lessen the mental and physical impairments caused by the disorder. Not only is there a demand for early detection, but also the public health system has moved toward educating citizens about the prevention of certain genetic conditions. Furthermore, there is an increasing interest in the services available to meet the needs of children with special health care concerns. Questions are asked whether the current services are appropriate, adequate, continuous, and effective. Families of children with special health care needs require diverse services – diagnostic, educational, medical, dental, and mental health services to name a few. The State of Nebraska currently has in place most of these services, however, the service agencies and providers lack a seamless collaborative relationship. Additionally, there is a lack of infrastructure available to connect these caregivers and supporting agencies.

The following is Nebraska's Plan for Newborn Screening and Genetics Services. The purpose of this plan is to guide the development of a system-wide coordination of care for children with special health care needs with genetic conditions. This begins with the Newborn Screening Program and culminates in the transitional services needed to prepare for adulthood. The goal is to create a seamless system that benefits all involved – patients, families, caregivers, health care professionals, and services providers.

We would like to thank our Advisory Committee members (Appendix A) who have graciously given of their time and talents. Without their efforts, this plan would still be an unwritten aspiration.

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This project was supported by Grant #5 H46 MC 00202-02 from the Maternal and Child Health Bureau (Title V, Social Security Act), Health Resources and Services Administration, Department of Health and Human Services.

List of Acronyms in this Document

AAP -- American Academy of Pediatrics
AAFP -- American Academy of Family Physicians
ACMG -- American College of Medical Genetics
ACOG -- American College of Obstetrics and Gynecology
AHRQ -- Association for Health Research Quality
AMCHP -- Association of Maternal and Child Health Professionals
AMA -- American Medical Association
APHL -- Association of Public Health Laboratories
ASTHO -- Association of State and Territorial Health Officers
CDC -- Centers for Disease Control
CONNECT -- Coordinating Options in Nebraska's Network through Effective Communications and Technology (an integrated computer tracking system)
CORN -- Council of Regional Networks for Genetic Services
CPH -- Congenital Primary Hypothyroidism
CSHCN -- Children with Special Health Care Needs
DD -- Developmental Disabilities
EBC -- Electronic Birth Certificate
EI -- Early Intervention
EIF -- Emergency Information Form
FAQ -- Frequently Asked Questions
HHSS -- Health and Human Services System
HIPAA -- Health Insurance Portability and Accountability Act of 1996
HRSA -- Health Resources and Services Administration
HRTW -- Healthy and Ready To Work
IDEA -- Individuals with Disabilities Education Act
LEND Project -- Leadership Education in Neurodevelopmental and Related Disabilities Project
MCAD -- Medium Chain Acyl-Co A Dehydrogenase deficiency (a metabolic disorder diagnosed by MS/MS)

MCH -- Maternal and Child Health
MCHB -- Maternal and Child Health Bureau
MHCP -- Medically Handicapped Children's Program (Nebraska's CSHCN Program)
MHPSA -- Mental Health Professional Shortage Area
MMI -- Munroe-Meyer Institute for Genetics and Rehabilitation (a UNMC center)
MOD -- March of Dimes
MS/MS -- Tandem Mass Spectrometry (an instrument used for testing for up to 30 or more disorders in an expanded newborn screen)
MUAs -- Medically Underserved Areas
MUPs -- Medically Underserved Populations
NBS -- Newborn Screen(ing)
NBSGAC -- Newborn Screening and Genetics Advisory Committee
NBSTAC -- Newborn Screening Technical Advisory Committee
NDE -- Nebraska Department of Education
NICU -- Neonatal Intensive Care Unit
NIH -- National Institutes of Health
NMA -- Nebraska Medical Association
NNHSP -- Nebraska Newborn Hearing Screening Program
NNSGRC -- National Newborn Screening and Genetics Resource Center
NNSP -- Nebraska Newborn Screening Program
OB/GYN -- Obstetrics and Gynecology
PKU -- Phenylketonuria (a metabolic disorder causing mental retardation if not treated)
Project LEARN -- Leadership Education in Advancements in Research in Neuroscience
PTIC -- Parent Training and Information Center
RFP -- Request for Proposals
TIPS -- Tracking Infant Progress Program
UNMC -- University of Nebraska Medical Center

Executive Summary

Up to twenty percent of children and youth have special health care needs (2010 Express). As with any other state, it is difficult in Nebraska to ascertain the exact number of children with special health care needs (CSHCN). The definition varies from program to program, and children using more than one service may be counted more than once. Other children may “fall through the cracks” because many of the service agencies in Nebraska lack integrated databases through which referrals and information sharing can be accomplished. Because of the primarily rural composition of Nebraska, the availability of health care services, particularly specialized health care, is limited for many of our citizens. Nebraska must assure not only the timely identification of children with special health care needs, but also the availability of well-coordinated, appropriate services.

Eastern Nebraska enjoys excellent genetics expertise through professionals in Omaha, Lincoln, and Fremont. Efforts are made to extend services statewide through clinics offered in greater Nebraska. However, many health care providers serving children with genetic conditions do not coordinate their services with other providers. A collaboratively developed state genetics plan is needed for a more systematic and cohesive delivery system. In July 2001, the Nebraska Health and Human Services System (HHSS) was awarded a federal grant for the project, “State Genetic Plan for Comprehensive Integrated Newborn Screening and Genetics Infrastructure for Children with Special Health Care Needs and Genetic Disorders.” The purpose of this project was to conduct a comprehensive needs assessment of genetic services in Nebraska resulting in a State Genetics Plan for newborn screening and infrastructure development.

The project established a Newborn Screening and Genetics Advisory Committee (NBSGAC) which defined its target population as children with special health care needs and genetic conditions who:

- 1) have been identified by the Newborn Screening Program, or
- 2) have a diagnosed genetic and/or congenital disorder(s), or
- 3) are at increased risk (as defined by positive family history or known carrier of a susceptibility gene) for a genetic and/or congenital disorder that may manifest within the childhood period (through age 21) which places them at increased risk for chronic physical, developmental, behavioral or emotional disorders, and who also require health and related services of a type or amount not usually required by children.

The major goal for the first year was to complete a comprehensive genetic services and newborn screening infrastructure needs assessment. This included reviewing existing legislation, service structures and resources. Additionally, the NBSGAC assessed whether children with special health care needs (CSHCN) and genetic conditions are identified at an early age and are receiving appropriate treatment and follow-up. In the meantime, issue-oriented work groups were organized around the topics of: database systems, finance, legislation, policy, and personnel. Following the collection and distribution of data, committee members assessed the information obtained in light of issues pertinent to their

work groups. The members then identified the strengths, weaknesses, and needs within the service delivery system relating to their topic.

The major strengths of the present system include:

- ☐ Existing or available program databases/warehouses
- ☐ Health related services
- ☐ NBS related services
- ☐ Family support services
- ☐ Tracking Infant Progress (TIPS) Program

NBSGAC members identified approximately seventy particular needs. The committee was able to group these under nine main headings:

- ☐ Parent Education
- ☐ Physician Education
- ☐ Desirable Database
- ☐ Legislation
- ☐ HIPAA/Informed Consent
- ☐ Medical Home
- ☐ Transitioning
- ☐ Policies/Procedures
- ☐ Expanded Services

These nine areas were further condensed into four primary areas of infrastructure needs, specifically:

- ☐ Policies and procedures
- ☐ Financial and legislative support
- ☐ Education
- ☐ Expanded services

In the second year, the committee developed the Newborn Screening and Genetics State Plan. The Plan allows for the provision of family focused, community-based, culturally competent, comprehensive genetic health care, outreach, intervention, and education integrating the public health and private sector arenas for children with special health care needs and genetic disorders. The needs identified in this assessment process were the basis for developing this plan.

The committee identified six core areas of action:

- ☐ Education of parents and healthcare providers
- ☐ Financial support to implement the recommended actions
- ☐ Enhanced staffing and staff training
- ☐ Legislation to establish long-term, sustainable funding sources with supporting policies and procedures
- ☐ An integrated, or linked database system
- ☐ Expanded services to establish a comprehensive CSHCN and Newborn Screening Program

With improved data capacity and integration of data systems, better measurement capacity should be realized. With the development of appropriate communication and service linkages, Nebraska should see an increase in the percent of CSHCNs with genetic

disorders who receive timely, quality, integrated, comprehensive, family centered, culturally appropriate services coordinated through a medical home.

A requisite element for successful implementation of these recommendations is the political will to make these issues **priorities**, now and in the future. In the current budgetary climate, a priority commitment becomes paramount to adequately serve children with special health care needs. The plan requires a commitment to:

1. Enabling legislation
2. Funding
3. Personnel

In summary, the committee recognizes the strengths in Nebraska's services for children with special health care needs with genetic conditions and the newborn screening program. However, many distinct needs are apparent. The committee's efforts have culminated in a plan to fill these gaps in services and create a more connected, thorough system for the children and their families.

Family Story #1

My husband and I became proud parents on April 17, 2002, when our first child, Evan Lee, was born. We went home from the hospital on Friday, a little nervous, as all first time parents are, and concerned. Evan was very jaundiced and already had a diaper rash. In addition to the cord care and the circumcision care, we were overwhelmed. We were having a rough time with my recovery, as well as with breastfeeding. All of these concerns, coupled with no sleep and daily lab draws for bilirubin levels, had us very anxious. On our third day home from the hospital, we received a call on our answering machine that Evan's newborn screen for PKU was abnormal and it was urgent that we call our pediatrician's office. Having been a nurse for four years, and currently working towards my Family Nurse Practitioner degree, I was a little familiar with PKU. I was under the impression that I would never see it in practice, especially in my own child, because it was so rare. I began to cry and think "this cannot be" and my husband was soon in tears saying, "What is it? What does it mean?" I did not know about current advances and treatment in PKU, and to me this possible diagnosis meant that my sweet son was going to have severe problems.



Newborn Patient with PKU

Our pediatrician called us back, letting us know he had seen positive screens that were not abnormal when checked again, and this gave us some confidence that there was a strong possibility Evan did not have PKU. We went to the hospital for a third time that day to have another screen for PKU, and although tearful, we felt we had to hope for the best.

On Monday, Children's Hospital Metabolic Clinic called, after we had just returned home from our bilirubin level, and said they would like us to be there in one hour to meet with Dr. Wiltse. Evan was only five days old at this time. I hung up and looked at my husband, wondering if Evan had PKU or if we were just going in to discuss what an elevated level could mean. Our pediatrician told us we would probably be going to the clinic to be evaluated, as all abnormal screens are. We packed up again, still hoping for the best.

When we got to the hospital, we were greeted by Dr. Wiltse in the waiting area and escorted to a back room, where several people were waiting. There was a dietitian, a social worker, several residents and students, and our family. It was in this room that we found out that Evan did have PKU.

That day was such a blur. I remember crying, while Dr. Wiltse tried to assure us that Evan would do everything that a "normal" child would do. It is just hard to believe this until you see it for yourself. They told us about the special formula and told us a little about the diet, but mainly that we would be working quite closely with a dietitian from Children's. They talked to us about lab draws and told us that one day, when we felt comfortable, we could begin drawing Evan's lab at home and mailing them in to NHS (Nebraska Health Systems laboratory). They also told us they would introduce us to other children with PKU so that we could see that Evan would grow up just fine. We left the hospital that day, still not believing that it could be true. Our perfect son and our perfect image of how life would be were not going to be that way. The clinic had given us several articles and papers to read about PKU, but one of the best things they gave us to get us through the first few days was a poem called "Holland"*. It made me feel that there was someone out there who knew exactly what I was feeling and that it was okay to feel that way. It also made us realize that although the situation was not as we had planned, it was still a wonderful experience and we needed to enjoy it and accept it, rather than wishing we were still heading towards our idealistic vision.

We had excellent support from Children's Hospital, especially Dr. Wiltse. He accepted our calls and answered our questions confidently. He sent us a short letter in the mail with some information he felt we

would be interested in. My husband pulled several stories of kids with PKU off the Internet so we could read them and feel confident that, yes, Evan would be okay. The more we read, the more thankful we were that we had quickly been informed of Evan's diagnosis of PKU and received treatment, as this is very important to prevent neurological damage. The same health professionals were at every one of our visits to the clinic, so we quickly began to feel comfortable and even look forward to our appointments.

We began to draw Evan's labs on our own when he was around three months old. The draws went smoothly some weeks, and not so good on others, but we have come a long way. Evan is now eight months old and developing beautifully. He is a strong willed, happy baby who we enjoy with all of our hearts. The measuring and weighing of all the foods, the adding of all the mg of PHE (phenylalanine), are all we have known, and seem like second nature to us. The diet has just begun, but we are doing great with it. We draw Evan's lab every week and then drive it to the hospital. We receive our results the next day, which makes us more comfortable with diet changes and accuracy of readings. Each week we talk to the dietitian and make any necessary changes, but the changes are minimal at this time.

There is a lot of added stress, especially in the newborn period, with added doctors visits, extra co-pays, buying scales and cookbooks and blood draw supplies. This is when we are thankful that Nebraska is so supportive of PKU care. I cannot imagine the financial strain of the disorder on top of dealing with the diagnosis itself, especially as new parents.

There is extra stress when you are trying to maintain breastfeeding in the beginning, pumping six times a day on top of feeding the baby every 2-3 hours. It is easy to feel like no one understands the intensity of this part, even new mothers. There are days when you look at your friends who have children and you think, "I wish I could just feed him Cheerios or breastfeed him whenever I wanted to". Or, you hear the song "take me out to the ballgame, buy me some peanuts and cracker jacks" and it just makes you sad. But then you make yourself think about how lucky you are to be able to spend your life with a healthy, beautiful child who simply needs a special diet, and those feelings disappear. Yes, it has been a very challenging time for us, but we have grown so much and appreciate the health we all have. We have met some wonderful people, and we appreciate the gift we have been given, a beautiful son. We are so thankful to Robert Guthrie for the discovery of PKU screening, and to the Nebraska Newborn Screening Program and as our son has been given the chance to live a long, productive life, just like any other child, because of a simple screen. It's amazing how your life can be forever changed in just a single moment, with a single test. We are so blessed. We are so thankful for the life we have been given and the chance for Evan to make the most of his gifts of health and life.

**** Welcome to Holland by Emily Pearl Kingsley***

I am often asked to describe the experience of raising a child with a disability - to try to help people who have not shared that unique experience to understand it, to imagine how it would feel. It's like this..

When you're going to have a baby, it's like planning a fabulous vacation trip - to Italy. You buy a bunch of guidebooks and make your wonderful plans. The Coliseum. The Michelangelo David. The gondolas in Venice. You may learn some handy phrases in Italian. It's all very exciting.

After months of eager participation, the day finally arrives. You pack your bags and off you go. Several hours later, the plane lands. The stewardess comes in and says, "Welcome to Holland."

"Holland!" you say. "What do you mean, Holland? I signed up for Italy! I'm supposed to be in Italy. All my life I've dreamed of going to Italy". But there's been a change in the flight plan. They've landed in Holland and there you must stay.

The important thing is that they haven't taken you to a horrible, disgusting, filthy place, full of pestilence, famine, and disease. It's just a different place. So you must go out and buy new guidebooks. And you must learn a whole new language. And you will meet a whole new group of people you would never have met.

It's just a difference place. It's slower-paced than Italy, less flashy than Italy. But after you've been there for a while and you catch your breath, you look around, and you begin to notice that Holland has windmills. Holland has tulips. Holland even has Rembrandts.

But everyone you know is busy coming and going from Italy, and they're all bragging about what a wonderful time they had there. And occasionally you might say to yourself, "Yes, that's where I was going. That's what I had planned."

Introduction

Up to twenty percent of children and youth have special health care needs (2010 Express). As with any other state, it is difficult in Nebraska to ascertain the exact number of children with special health care needs (CSHCN). The definition varies from program to program, and children using more than one service may be counted more than once. Other children may “fall through the cracks” because many of the service agencies in Nebraska lack an integrated database through which referrals and information sharing can be accomplished. Because of the primarily rural composition of Nebraska, the availability of health care services, particularly specialized health care, is limited for many of our citizens. Nebraska must assure not only the timely identification of children with special health care needs, but also the availability of well-coordinated, appropriate services.

In Nebraska, multiple programs and data systems are in place for public health purposes that share some common elements, yet are not integrated in any fashion. Data are generated multiple times in different formats for different purposes. For example, medical records personnel at the local hospital may enter birth certificate and birth defects data, while a postpartum nurse completes demographic data for newborn screening, and an early intervention specialist from the educational service unit records developmental evaluation and test results.

The most obvious example of this fragmented system is illustrated in the Nebraska Newborn Screening Program. Speedy and accurate communication is vital to assure that newborns are screened and, if positive results are found, that they receive follow-up testing and all needed services as quickly as possible. As the scope of screening increases, the challenges of timely and accurate intervention increase exponentially. The current NBS database system has two major problems. First, it does not allow staff to track newborns through the screening process. Second, the database is extremely limited in its usefulness to partners in the program, particularly physicians. Currently, the newborn screening program struggles to keep physicians across the state educated about the protocols for screening. It is also a challenge to inform physicians of the resources available to parents when positive results are found, particularly with regard to the optional supplemental screening. In addition, many other providers of health care and related services may eventually be part of the child's life, all of whom have separate data systems. Linking these disparate data systems would make it easier for primary care physicians to serve as care coordinators, thus increasing the likelihood that families will be able to navigate the systems successfully.

Background on the State Plan Project

Eastern Nebraska enjoys excellent genetics expertise through professionals in Omaha, Lincoln, and Fremont, and efforts are made to extend services statewide. Other specialty clinics and care programs offer services to specific subpopulations. However, these entities lack a cohesiveness that can best be developed through a collaborative effort in a state genetics plan. As in the previous example, despite improvements in Nebraska's Newborn Screening Program over the last decade, certain components of infrastructure continue to be weak or absent, including:

- 1) Lack of a state genetics plan for newborn screening

- 2) Under-representation of minorities, consumers, the general public, private sector interests, related public health agencies, and a medical ethicist on the Newborn Screening Technical Advisory Committee (NBSTAC)
- 3) Poor integration of the newborn screening system with the health care delivery system
- 4) A multi-laboratory system functioning without benefit of contractual or regulatory authority for quality assurance
- 5) Lack of cost containment for laboratory charges
- 6) Lack of stable funding sources for the non-lab components of newborn screening
- 7) Lack of a long-term follow-up system (infrastructure for surveillance and research)
- 8) Lack of integration of newborn screening data with other points of early identification, such as the birth defects registry
- 9) A data system that does not allow retrieval of information from the state newborn screening program by the private sector primary care providers; and
- 10) Collaborative efforts but little integration among public health agencies and private sector providers;
- 11) Lack of newborn screening data integration across state lines.

Some of the infrastructure needs have been altered with the passage of newborn screening-related legislation in 2001 (LB 235). This law mandated expanded screening for Medium Chain Acyl Co-A Dehydrogenase Deficiency (MCAD). Furthermore, the law authorized the Nebraska Health and Human Services System (HHSS) to contract with a single lab to perform all of Nebraska's newborn screening testing, and provided a fee to partially cover the non-laboratory components of the screening. However, with the expanded screening has come a need for greater consumer and provider education; a method of consent/dissent for results generated by supplemental screening beyond MCAD; development and implementation of a pilot study; resource development; and expansion of communication/data systems to accommodate additional screening results.

In July 2001, the Nebraska Health and Human Services System was awarded a federal grant for the project, "State Genetic Plan for Comprehensive Integrated Newborn Screening and Genetics Infrastructure for Children with Special Health Care Needs and Genetic Disorders." The purpose of this project was to conduct a comprehensive needs assessment of genetic services in Nebraska resulting in a State Genetics Plan for newborn screening and infrastructure development. To create the state plan, the Nebraska Newborn Screening and Genetics Advisory Committee (NBSGAC) was formed. Five work groups from this committee met to conduct the work of completing the needs assessment and plan. The following documents provided the framework for these tasks: the American Academy of Pediatrics Newborn Screening Task Force Report "Serving the Family from Birth to the Medical Home", the Council of Regional Networks for Genetic Services (CORN) "Guidelines for Clinical Genetic Services for the Public's Health", and "National Agenda for Children with Special Health Care Needs, Healthy People 2010". The needs assessment and plan address:

- ☐ integration of the newborn genetic screening program with other points of early identification of children with genetic conditions and special health needs
- ☐ integration of early identification systems and early intervention systems
- ☐ retrieval of information from the newborn genetic screening programs by private sector primary care providers, and

- ❑ facilitation of collaborative efforts among public health agencies and private sector providers concerning newborn genetic screening

The major goal for the first year was to complete a comprehensive genetic services and newborn screening infrastructure needs assessment. This included reviewing the existing legislation, services structures and resources. Additionally, the NBSGAC assessed whether children with special health care needs and genetic conditions are identified at an early age and are receiving appropriate treatment and follow-up. In the meantime, issue-oriented work groups were organized around the topics of: database systems, finance, legislation, policy, and personnel.

Data was researched and gathered, then made available to the Advisory Committee to evaluate the existing infrastructure. Information for the work groups was obtained from a variety of sources including, but not limited to: journal articles, existing program budgets and policies, other state newborn screening programs, Internet sites, personal communications, and surveys of parents and physicians who have had experience with the NE Newborn Screening Program (NNSP). The results of those surveys are found in Appendices B, C, and D. Additionally, the "*State Assessment Tool*, Comparing State Genetics Services to the CORN Guidelines for Clinical Genetic Services for the Public's Health" was completed by the Project Director and Principal Investigator to assist with the needs assessment process (Appendix E). Following the collection and distribution of data, committee members assessed the information obtained in light of issues pertinent to their work groups. The members then identified the strengths, weaknesses, and needs within the service delivery system relating to their topic.

In the second year, the committee developed the Newborn Screening and Genetics State Plan. The Plan allows for the provision of family focused, community-based, culturally competent, comprehensive genetic health care, outreach, intervention, and education integrating the public health and private sector arenas for children with special health care needs and genetic disorders. The needs identified in this assessment process were the basis for developing this plan.

Another primary process goal for the planning grant was to improve collaboration among public health agencies, private providers and consumers of services for CSHCN. The measurement of this goal has been accomplished by a survey of the Advisory Committee members. The survey documented committee member's perceptions of inter-agency relationships, and actual collaborative activities and projects. This survey was provided prior to the first NBSGAC meeting and at the end of Year One, and will also be given prior to the end of Year Two. The current results are available in Appendix F.

Composition of the Advisory Committee

The needs assessment task was carried out by the Newborn Screening and Genetics Advisory Committee (NBSGAC). Expansion of the previously established Newborn Screening Technical Advisory Committee (NBSTAC) into the NBSGAC for this project included representation from:

- ❑ ***Pediatric specialties*** – genetics, endocrinology, hematology, pulmonology; *State Agencies* – Office of Minority Health and Human Services, Medicaid/Medicaid Managed

Care Program, the state's co-lead agencies (HHSS and Dept. of Education) for Early Intervention (Part C of IDEA), Childhood Immunization Program, Childhood Lead Poisoning Program, Nebraska's Vital Records and Data Management systems, State Information Systems and Technology Services, Medically Handicapped Children's Program, Nebraska Newborn Screening Program, and the Nebraska Newborn Hearing Screening Program

- ❑ **Medical Academies** - Nebraska Chapters of the American Academy of Pediatrics (AAP) and American Academy of Family Physicians (AAFP) (representing the medical home)
- ❑ **Consumers and Consumer Groups** - representing metabolic, endocrine, and hematologic disorders, March of Dimes Nebraska Chapter, Sickle Cell Anemia Foundation, and the Parent Training and Information (PTI) Center
- ❑ **State Legislature**
- ❑ **The Nebraska Hospital Association**
- ❑ **The Nebraska Medical Association**
- ❑ **And, a medical ethicist**

(For a complete listing of members, please see Appendix A) The NBSGAC was further divided into five work groups: Data Infrastructure, Financing, Legislative, Personnel Resources, and Policy.

Definition of CSHCN

In order to complete the needs assessment process, the NBSGAC needed to define the population of CSHCN that the project would address. The committee agreed upon a non-categorical, functional definition of children with special health care needs with genetic conditions. These are children who:

- 1) Have been identified by the Newborn Screening Program, or
- 2) Have a diagnosed genetic and/or congenital disorder(s), or
- 3) Are at increased risk (as defined by positive family history or known carrier of a susceptibility gene) for a genetic and/or congenital disorder that may manifest within the childhood period (through age 21) which places them at increased risk for chronic physical, developmental, behavioral or emotional disorders, and who also require health and related services of a type or amount not usually required by children.

Current Strengths In the Newborn Screening and Children with Special Health Care Needs with Genetic Disorders System

The current strengths in the newborn screening infrastructure and for CSHCN with genetic disorders service system were clustered into five primary areas.

- ❖ The **existing or available program databases/warehouses** currently collect a large number of data elements through various program and service databases. For example, the CONNECT data warehouse system offers many advantages such as the ability to identify what services a child/family has been referred to, which services they are receiving, what services are needed but not available, and which services were declined. The CONNECT computer system is an integrated database (between HHS and NDE) which includes the Medically Handicapped Children's Program, Early Development Network (Early Intervention), Aged and Disabled Waiver (including eligible children), and the Lifespan Respite Subsidy Program (which includes children).

The children and youth served by the Disabled Persons and Family Support Program are also included in this database.

- ❖ The **health related services** for CSHCN in Nebraska are provided by a solid core of skilled professionals and the current funding sources are diverse. Close collaboration between the Munroe-Meyer Institute at the University Nebraska Medical Center (MMI/UNMC), HHSS, and NDE have allowed for the maximum use of funds and professionals. The multiply trained specialists allow coupling of many services and provision of a large number of services by the existing staff. Nebraska has eight board certified or board eligible genetic counselors, a reasonable number for the size of the state. Prior to the state budget cuts in 2002, quarterly outreach developmental/genetics clinics for CSHCN were conducted in Scottsbluff, North Platte, Kearney, Hastings, and Norfolk and included genetic counseling. In addition, two clinics a year were held in Grand Island. However, as of October 2002, the clinics will only be held at Scottsbluff and Kearney; and as of October 2003, the availability of outreach clinics is uncertain.
- ❖ The **NBS related services** were noted for dedicated staff, efficient turn around time and excellent follow-up and tracking. There is a strong working relationship between the NBS staff, Technical Advisory Committee, labs, specialists, and MMI/UNMC. The NBS Practitioner's Manual is thorough and provides informative web sites. Another strength of the program is that the state pays for the medical foods and formula needed by some children and adults identified with metabolic disorders through newborn screening.
- ❖ **Family support services** were acknowledged for the willingness of previously identified families of CSHCN to connect with the parents of newly diagnosed children. These families have a wealth of experience and resources, and are willing to share this. Additionally, established Parent-to-Parent Networks serve as helpful resources.
- ❖ The **Tracking Infant Progress (TIPS) Program** identifies and tracks infants who have been admitted to the Neonatal Intensive Care Unit following delivery. This allows for certain CSHCN to be identified at an earlier age and referred to appropriate services. As a statewide program, it provides referral services for both rural and urban families through a variety of locations.

Summary of the Needs Assessment

NBSGAC members identified approximately seventy particular needs grouped under nine main headings: Parent Education, Physician Education, Desirable Database, Legislation, HIPAA/Informed Consent, Medical Home, Transitioning, Policies/Procedures, and Expanded Services. These nine areas were further condensed into four overriding areas of infrastructure needs, specifically: policies and procedures, financial and legislative support, education, and expanded services.

- ❖ **Policies and procedures** are necessary for appropriately funding and staffing the Nebraska Newborn Screening Program providing quality assurance of all components of the programs and data collection, following applicable federal guidelines such as HIPAA, and maintaining a database and data sharing system.
- ❖ **Financial and legislative support** is required to implement the recommendations in the State Plan resulting from this needs assessment. An earmarked, stable funding source for the NNSP, and a stable multi-funding source for the CSHCN service system is necessary to provide for continual modernization and maintenance of services.

- ❖ The ***education of both parents and healthcare providers/physicians*** was identified as an essential need. Increased education, educational tools, and continuing education were deemed vital to assure communication of medical information, prevent delays in patient care, and increase the comfort and confidence of both parents and physicians.
- ❖ The need for ***expanded transitional services, long-term care services, and medical home implementation*** was expressed for creating a comprehensive and complete CSHCN and Newborn Screening Program.

Summary of the Recommended Actions

After reviewing the necessary information, the committee concluded there are six core areas of action that could address the identified needs:

- ❖ ***Education*** of both parents and healthcare providers/physicians was identified as an important need. Increased education, educational tools, and continuing education were deemed vital to assuring communication of medical information and prevention or delay in patient care, as well as an increase in the comfort and confidence of both parents and physicians.
- ❖ ***Financial support*** is required to implement the recommended actions in the State Plan resulting from this needs assessment. As previously mentioned, an ear-marked, self-supporting fee structure for the NNSP, and a stable multi-funding source for the CSHCN service system is necessary to provide for continual updating and maintenance of services.
- ❖ Additional, or more adequately trained ***personnel*** are crucial to providing the appropriate care for CSHCN and for staffing the many programs that support both the NNSP and CSHCN services.
- ❖ ***Legislative action*** is necessary for creating long-term, sustainable funding sources for the Newborn Screening Program and adequate genetics services. In response to legislation, policies and procedures are required to: appropriately staff the NNSP program; to provide quality assurance of all components of the programs and data collection; to follow applicable federal guidelines such as HIPAA; and to maintain a database and data sharing system.
- ❖ Many of the services for CSHCN would be best coordinated through an integrated, or linked ***database system***, which would be available to those services and providers granted permission by the patients and/or parents.
- ❖ Finally, a need for ***expanded services***, especially those in transitioning, long-term care, and medical home implementation was expressed for creating a comprehensive and complete CSHCN and Newborn Screening Program.

State Plan Summary

A requisite element for successful implementation of these recommendations is the political will to make these issues ***priorities***, now and in the future. In the current budgetary climate, a priority commitment becomes paramount to adequately serve children with special health care needs. The plan requires a commitment to *enabling legislation, funding, and personnel*. In summary, the committee recognizes the strengths in Nebraska's services for children with special health care needs with genetic conditions and the newborn screening program. However, many distinct needs are apparent. The committee's efforts have culminated in a plan to fill these gaps in services and create a more connected, thorough system for the children and their families.

Nebraska Demographics



Nebraska is a geographically large state, covering over 77,000 square miles. The state is located in the east-central area of the Great Plains. It is bordered by South Dakota to the north, Wyoming and Colorado to the west, Kansas to the south, and Missouri and Iowa to the east. Nebraska is about 390 miles from west to east, and 210 miles wide. It is the 16th largest state geographically, with the 38th largest population. According to the 2000 U.S. Census, the total population in Nebraska was 1,711,263, with the median age of the population being 35.3 years. Although Nebraska is considered a rural state, the rural populations continue to dwindle, while the larger cities continue to grow. Nearly half of Nebraska's population lives in the three counties – Douglas, Lancaster and Sarpy, which includes Omaha, the largest city, and the capital city of Lincoln. Douglas County alone has more than a quarter of the state's population. (Nebraska Blue Book) There were approximately 25,500 live births in 2002 at 69 birthing hospitals. The vast majority of hospitals are located in counties classified as rural or small urban with each hospital averaging less than 50 births per year. However, the majority of births occur in the ten urban hospitals found in Nebraska's largest cities, Lincoln and Omaha.

Nebraska's large land expanse creates unique health service delivery issues. The state is sparsely populated by national standards, with 32 of its 93 counties designated as frontier counties (with six or fewer individuals per square mile). Although Nebraska's total population has grown considerably during the 1990s, many small rural counties that are not near a regional economic or health center continue to decrease in size. Most of the decrease in these counties resulted from out-migration of the younger population (18 to 45 years). Smaller population bases make it more difficult to recruit and retain physicians and other health care professionals. A small population base also makes it more difficult to operate institutional services, such as hospitals, and finance other types of services such as mental health, public health, emergency medical services, and long-term care services.

Nebraska's population is becoming more racially and ethnically diverse. Since 1990, the state has seen more growth in minority populations, particularly Asians and Hispanics. Hispanics, the fastest growing minority population, are now the largest minority group in

the state. (Nebraska Blue Book) Since 1990, the Hispanic population has increased by 94.9%. The Asian American population has grown by 71%, the African American population has grown 15.7%, while the Native American population has grown 15.3%, and the white population has grown only 4.1%. According to the 2000 U.S. Census, 86% percent of the population described themselves as white, 5.5% were Hispanic or Latino, 4% were Black, 1.3% were Asian, and 0.9% were American Indian or Alaska Native.

Since the mid 1990s, Nebraska has become a center of relocation for refugees from many countries. For example, Sudanese refugees resettled in states across the country, and have relocated to Nebraska in such numbers that Nebraska has the highest number of Sudanese in the country. The size of the racial/ethnic minority population in Nebraska is thought to be underestimated, particularly for Hispanic Americans in the state. The mobility of this segment of the population, particularly migrant workers and their families, as well as the presence of an undetermined number of undocumented workers makes it difficult to arrive at an accurate number. Likewise, the number of Asian Americans in Nebraska may also be underestimated to some extent, as a result of the influx of recent immigrants. (Health Status of Racial and Ethnic Minorities in Nebraska)

In general, the minority population tends to be younger, have lower incomes, higher poverty, and less insurance coverage. They are also more likely to be employed in high-risk occupations such as meat packing plants and farm labor. As a result, these population groups often experience difficulty gaining timely access to health and medical services. Even when services are available, language and cultural barriers may prevent effective utilization of these services. The diversity in languages can be demonstrated by looking at the number of countries and number of languages represented in the Lincoln Public School District (2nd largest metropolitan area). During the 2002/2003 school year, 42 languages from 53 countries were spoken. A language other than English is spoken in 7.9 % of homes (4.9 % Spanish, 1.7 % Indo-European, 0.9 % Asian and Pacific Island).

The median household income was \$39,250 in 1999; 6.7 % of families were considered to have "poverty status." Although the state is largely based on agricultural industries, only 1.6% of the population state that their occupation is farming, fishing, or forestry.

Demographics and geography hinder Nebraska's direct health care services. Half of the population is located in five counties in the state, where most of the health services are also located; however, geographically, most of Nebraska has been federally designated as Health Professional Shortage Areas (HPSAs), Medically Under-served Areas (MUAs), and Medically Under-served Populations (MUPs).

In 2001, over a third (33/93) of Nebraska's counties were designated, either in full or in part, as primary care HPSAs. These shortage areas potentially affect access to primary care health providers for more than 25% of Nebraska's population. Based on 2000 Census estimates, more than 12% of the state's population lives directly within a HPSA. In addition, 73 of Nebraska's 93 counties have been designated, in full or in part, as containing MUAs or MUPs. Over 22% of the state's population live within the designated areas and are potentially affected by a shortage of health services.

Within state designated HPSAs, a high degree of shortage exists in each of the defined health specializations, including: family practice, general surgery, internal medicine, pediatrics, OB/GYN, psychiatry, dentistry, pharmacy, occupational therapy, and physical therapy. Three-fourths of Nebraska's counties currently have a shortage of family practice physicians (70/93), 91% have a shortage of pediatricians (85/93), and 91% have a shortage of OB/GYNs (85/93). Of the 130 pediatric specialists (representing 26 different specialty areas) in Nebraska, all but 23 are located in Douglas County. All but six pediatric specialists practice in the eastern part of the state. In addition to the 130 pediatric specialists, Nebraska has 155 pediatricians (total 285) located in only 15 of the states' 93 counties. In 2001, the U.S. Department of Health and Human Services designated over 70% of Nebraska's counties (66/93) as Mental Health Professional Shortage Areas (MHPSAs). Additionally, the populations of nearly half of Nebraska's counties were affected, to some extent, by a lack of access to dental care. Forty-three counties were designated as State Dental Shortage Areas. (NE Health Information Project 2001 Databook) The availability of medical specialty services varies and tends to exist only in the more urban areas of Nebraska. In many communities, newborns requiring referrals, or evaluation from pediatricians and sub-specialists, face issues of access to appropriate services.

As stressed in the "National Agenda for Children with Special Health Care Needs, Healthy People 2010", every child in America should have a medical home. In other words, every child should have a primary health care provider who knows the child individually, cares for him/her, is aware of his/her needs, and is capable of providing culturally sensitive and appropriate medical services. According to synthetic* estimates, Nebraska has 78% of the CSHCN population with medical homes. (*Based on 1994 National Health Information Service data and on 1996 Child Protective Services data). And, 94% of Nebraska's CSHCN were covered either by private insurance or Medicaid (3006/3198) in 1999, according to the Medically Handicapped Children's Program (MHCP).

The 2001 Annual Report/2003 Application for MCH Title V funding stated that 2,584 children with special health care needs were served through the Medically Handicapped Children's Program in fiscal year 2001. Another 4,117 CSHCNs were served through other community-based subgrantees for a total of 6,701; the majority (58.1%) the majority of which were covered by Medicaid. The MHCP is involved in a number of projects aimed at, among other things, increasing the number of CSHCN who have a medical home and who are covered by some type of health care insurance. This work is achieved through a variety of collaborations with UNMC, Creighton University, Boys Town National Research Institute, Vocational Rehabilitation, Department of Education, Special Education, HHSS programs including Medicaid and Developmental Disabilities, families, advocates and local medical and education professionals throughout the state. These collaborations are further described in "Current NBS, CSHCN, and Genetics Programs."

Family Story #2

I have spoken to many families in the last three years about their experiences of having a child with a genetic disorder. After hearing some of their experiences with the initial diagnosis, how they weren't given any information or had a doctor who never heard of Wolf-Hirshhorn syndrome (WHS), I feel very fortunate that I lived in Omaha, NE.

When I was pregnant with Jamie, I always had a feeling that something just wasn't right. I told my OB several times that I felt something was wrong but our first ultrasound looked good. The early screening test came back okay and all my vitals etc were fine. It was my second ultrasound (6 month) that was the first sign that all might not be normal. The ultrasound tech measured Jamie and found that she didn't seem to be as big as she should have been. This was different than our first two girls as they were born 9+ pounds. I went to see a specialist and Dr. Foley couldn't find anything really wrong, just maybe some restriction in the flow in the cord. My OB suggested that we do an amnio and see if the lungs are mature enough to deliver so we could get a better idea on what was going on. My amnio results came back that the lungs were not mature so they ran the usual test to see if she had downs or trisomy 18. All came back okay. I remember my OB said that they can test every chromosome but he didn't see the need for it. So I was 38 weeks along and labor was induced. When Jamie was born I knew instantly that something wasn't right. She looked very different from my other two girls and she was basically skin over bones. She had all the facial features of a child with some kind of syndrome. She also had a cleft palate. We didn't know what to think. I have to say that on the outside we were calm but inside or when my husband and I were alone we were devastated. They transferred her to Children's Hospital where a geneticist named Dr. Olney saw her when she was 3 days old. Just from all Jamie's facial features and other characteristics (small hands, feet, dimple, head shape, flat nose etc.) Dr. Olney thought Jamie was 4p- or Wolf-Hirshhorn Syndrome (WHS). She ordered a blood test and sure enough Jamie had a deletion on her fourth chromo and had an official diagnosis of WHS.

I remember the day so well! Our doctor called to see if we could have a conference with all the doctors so they could give us Jamie's diagnosis. In our conference we had the geneticist, the genetic counselor, a social worker, and several pediatric specialists. Dr. Olney gave us the news and of course we were in shock. I think what helped us not to just break down and cry and lose it all together was that Crystal, the genetic counselor had a lot of information for us. She had parents of WHS children's names. She had info on the 4p- support group, which also has a listserv of parents of 4p- kids that has turned out to be an awesome, awesome place of knowledge for us. She had information on local support groups, Pilot Parents and Prism meetings. The social worker had all the information on early intervention services, contact information for a service coordinator who could help us with respite care etc. While I was overwhelmed at the time, I can't imagine what it would have been like for someone to give us this news and then send us home. A WHS baby born recently in another state made me appreciate all the resources I have here in Omaha. This poor family went thru so much. It was weeks before they even had a genetic test done and then when they got the results back, the doctor and the nurses had never even heard of this genetic condition. They had to go on the Internet and do research. Because we were so well informed - Jamie started OT/PT services at 3 months old. She has a teacher who helps us with developmental issues. We were able to have nurses come and watch Jamie and we could go out and enjoy ourselves, knowing that if Jamie has a seizure or medical emergency she is in good hands. Because I was able to meet and talk with other

WHS parents locally and nationally, I have been able to prevent many medical complications. For instance, parents told us about seizures and that they usually start around 7 to 8 months old - sure enough - when Jamie was 8 months she had her first grand mal seizure. Because we were prepared, we had diastat on hand and were able to give her an anti-seize med and by the time the paramedics arrived the seizure had stopped. I feel very fortunate that we have been lucky enough to have genetic counselors. I was able to call and ask questions that I couldn't ask another parent or felt like it was a dumb question and didn't want people to know that I was asking it. I have always felt good about the fact that if at anytime I have a question regarding Jamie, or need a resource for information, I can always call the office, Children's Developmental Clinic or Cranial Facial clinic.

Having a child with WHS has affected our whole family. We led a very active lifestyle before Jamie was born. We did a lot of camping and boating. While we have been able to still do those things we have had to be sure it's not too hot for Jamie. We have to take all our medical supplies and always make sure we know where the nearest hospital is. We learned a whole new appreciation for the little things in life. I remember Dr. Olney saying that it wasn't going to be easy and that we would have a tough road ahead of us. While those words were true we have found out that we are really strong people. Jamie has brought the best out of every single person she meets and that knows her. We didn't know that so many people cared for us until we had Jamie. While the journey has been very difficult at times, it has also been very enlightening and very rewarding.

Newborn Screening, Children with Special Health Care Needs, and Genetics Programs and Activities

Nebraska has a long history of a wide variety of programs and services focused on children with genetic disorders and special health care needs. There are also a number of data systems in place for tracking, monitoring and surveillance, few of which however are currently linked or integrated. The many years of collaboration between and among public health agencies, the state's university system, and private providers has evolved to a refined vision of services that are comprehensive, family-focused, community-based, and culturally competent in order to effectively meet the needs of families with children with genetic conditions and special health care needs. Unfortunately, the relationships are not always stable as funding sources fluctuate and the inability to access patient information from multiple service points hinders the continuity of care.

Nebraska's commitment to improved coordinated care is demonstrated by the restructuring of five separate agencies (Departments of Health, Social Services, Public Institutions, and Aging, and the Offices of Juvenile Services) into one integrated Health and Human Services System in 1997. This effectively brought programs relevant to services for children with special health care needs and genetic conditions that were previously separated administratively, into a single unified system. This included units such as the Office of Family Health (Title V/MCH Block Grant administrative unit), the Office of Minority Health, the Office of Aging and Disabilities, and the Developmental Disabilities System, as well as programs such as the Nebraska Newborn Screening Program (NNSP), Immunizations, Medically Handicapped Children's Program (MHCP, Nebraska's CSHCN program), Medicaid, Early Development Network (Part C, IDEA), Childhood Lead Screening, Birth Defects Registry, Electronic Birth Certificate (EBC), and Nebraska Newborn Hearing Screening Program (NNHSP).

A statewide system for genetics services in Nebraska was initiated over 24 years ago. This system is coordinated by the University of Nebraska Medical Center's (UNMC) Munroe-Meyer Institute for Rehabilitation and Genetics (MMI) and provides a comprehensive system for providing coordinated services for children with special health care needs. The current system includes professional education and technical assistance to community providers in the area of human genetics, developmental disabilities and neurobehavioral disorders. There is also a comprehensive environmental and exposure services system, and specialty clinics for consultation and coordination. Specialty clinics include:

- ☐ Cleft Palate and Craniofacial Clinic;
- ☐ Endocrine Clinic;
- ☐ Hemoglobinopathy Clinic;
- ☐ Metabolic Clinic;
- ☐ Midline Neurological Defects Clinic; and
- ☐ Specialty Clinics for Children and Youth.

Another existing clinic in Nebraska, with potential to be connected to the genetics system, is the Cystic Fibrosis clinic held in Omaha, Columbus, Norfolk, and North Platte.

Interdisciplinary clinics available across the state incorporate behavioral pediatrics, developmental pediatrics, and pediatric endocrinology. Many of these activities are supported through contract and sub-grants through partnerships with State programs such as the Medically Handicapped Children's Program (MHCP), Title V and other agencies such as the March of Dimes. The UNMC has demonstrated commitment to the core functions of public health by actively collaborating for many years with the Newborn Screening Program, Birth Defects Registry, Medically Handicapped Children's Program, and Newborn Hearing Screening for assessment, policy development, and assurance. The University's involvement has positively impacted Nebraska's population of children with special health care needs in all areas of the Maternal and Child Health pyramid. This involvement includes building infrastructure, participating in population-based services, enabling and directing health care services.

Nebraska is fortunate to have a wealth of health care professionals specializing in genetics. However, as with most medical specialties, the majority of the professionals are located in Omaha. The MMI houses three medical geneticists who are multiply trained in genetics, endocrinology, and metabolism and another equally trained physician is affiliated with the Children's Hospital. In Omaha, clinical general genetics services are provided by four medical geneticists and seven genetic counselors (four full-time, three part-time) at MMI, one medical geneticist at the Children's Hospital, and a medical geneticist and genetic counselor at Creighton University provide cancer genetics services. Two medical geneticists, one in Lincoln and the other in Fremont, provide part-time genetic services. Additionally, there are two PhD clinical geneticists, three cytogeneticists, and twenty cytogenetic technologists at MMI. The part-time geneticist in Lincoln also provides cytogenetic services. There are a total of six metabolic specialists (none full-time) in Omaha, as well as four full-time and two back-up pediatric endocrinologists. Obviously, the need in Nebraska is not for qualified health professionals, but for a permanently funded, coordinated, out-reach program of services.

The following are descriptions of the major programs and activities (past and current) related to newborn screening, genetics, and children with special health care needs:

Nebraska Newborn Screening Program

The Nebraska Newborn Screening Program (NNSP) screens for five mandated disorders: biotinidase deficiency (about 1:25,000), congenital primary hypothyroidism (about 1:6,000), galactosemia (about 1:80,000), hemoglobinopathies (about 1:600), and phenylketonuria (PKU) (about 1:12,000). In 2001, a handful of hospitals (34% of Nebraska births) were offering supplemental screening for 30-plus additional diseases. Beginning July 2002, MCAD was added to the mandatory screening panel. A total of 25,465 (99.8%) infants were screened for these disorders in 2002. There were 97 home births in 2002 and the NNSP continued community collaborations in order to facilitate the screening of these births. Infants diagnosed as positive for the following disorders were detected and facilitated into treatment through newborn screening: 2 infants with partial (treated) biotinidase deficiency; 15 infants with congenital primary hypothyroidism; 4 infants with sickle cell disease and two with sickle hemoglobin-C disease; one with phenylketonuria; and one with MCAD. Currently, two screening labs are used for five of the disorders (not MCAD). As of July 2002, 55% of births were receiving the supplemental screening through an out-of-state laboratory. Although the Nebraska Newborn Screening Program has no means to track the additional

diseases identified by the supplemental screen, nor the legal ability to facilitate follow-up on positively screened babies, the program is facilitating follow-up (since July 2002) on positive screens that are reported to them.

LB 432, passed in 2001, required development of regulations on dried blood spot storage, use and disposal. This legislation also required the NNSP to develop a model informed consent form for predictive and presymptomatic genetic testing. The NNSP promoted within the Department and to a Senator to sponsor introduction of LB 235 which would enable the program to contract with a single laboratory to do all of Nebraska's newborn screening testing. LB 235 would also expand the screening panel to include MCAD (Medium Chain Acyl CoA Dehydrogenase Deficiency), increase the newborn screening fee from \$3 to \$10 (supports the special foods and formula), add authority to the Department to require laboratory quality assurance participation, and reduce the overall cost of newborn screening. This bill was passed during the 2002 session and will be fully implemented by July 2003. When screening for MCAD, 20-30+ more disorders of fatty acid, amino acid, and organic acid may be included. Parents must consent to optionally screen for these 20-30 additional disorders.

As mentioned above, LB 235 focuses on expanding the existing program by revising statutory language to allow HHSS to accept competitive bids through an RFP process, and increases fees from \$3 to \$10. Not only would this allow the NNSP to reduce costs (by conducting all the testing at a single laboratory having the volume to reduce cost, and resources to ensure quality), but also quality assurance would be built into an annual contract thereby giving HHSS enforcement authority to change vendors should quality be less than optimal. With this new law, there is now enforcement authority for laboratories to comply with the regulations. Currently laboratories charge hospitals (who pass this on to the family/third party payer), \$53 to \$54.60. Of this fee, \$3 per infant goes to the state to support PKU foods and formula, \$50-51.60 is solely for the laboratory testing.

Nebraska Newborn Hearing Screening Program

The Nebraska Newborn Hearing Screening Program (NNHSP) coordinator is administratively under the direct supervision of the NNSP Manager. Newborn Hearing Screening is in its infancy in Nebraska, with statutory provision passed in 2000. Linkage with genetic services is seen by the Nebraska Newborn Hearing Screening Advisory Committee to be integral to serving children identified with hearing loss. Genetic referral protocols developed by the American College of Medical Genetics (ACMG) are to be reviewed by the NNHSP Advisory Committee and adapted, then adopted, by the system. An integrated database is being developed with HHS Information Services and Technology to facilitate tracking, monitoring, and follow-up for newborn screening and newborn hearing screening.

At the beginning of 2002, 24 hospitals were performing newborn hearing screening; by the end of the year, 57 hospitals were participating in newborn hearing screening. It is projected that 68 hospitals will be able to provide newborn hearing screening by the end of 2003. The preliminary data for 2002 shows that 92% of parents were given educational materials that recommended a hearing test, but that only 89% of the 25,509 of newborns received a hearing test during their birth admission. Nonetheless, this is greatly increased over the 61% of newborns who were screened in 2001. In 2002, almost 94% of the 22,665 receiving hearing screens, passed. Repeat screening was given to 596 newborns/infants, and

176 received diagnostic testing. Fifty-five newborns/infants (0.2%) were diagnosed with a hearing loss based upon a follow-up hearing test. This correlates with estimated frequency of deafness in childhood, which is as high as 1 in 500 (Mehl AL, Thomson V).

Some reasons why 100% of newborns are not receiving newborn hearing screening may include: smaller hospitals may not have the necessary screening equipment, one of the larger hospitals in Greater Nebraska started their newborn hearing screening in January 2003 (700 births per year), and primary care providers may not understand the value of early intervention to newborns with hearing loss at less than one year of age. In addition, without a centralized tracking system, the NNSP cannot assure that all infants are being tested.

If by December 2003, less than 95% of newborns are being screened, regulations will need to be promulgated to "require" hearing screening, and this will authorize the central tracking system to obtain data on every newborn, not just those failing the screen or not receiving a complete hearing screen.

Newborn Screening Technical Advisory Committee

The Newborn Screening Technical Advisory Committee has been in existence since 1993. The team has provided leadership and guidance on numerous issues including: expanding the newborn screening panel, developing and implementing regulations standardizing newborn screening practices, endorsing plans for the Nebraska Newborn Screening Program's electronic database system, and providing ongoing oversight of all components of the system including patient and provider education, laboratory testing, follow-up and treatment and quality assurance. This committee has and continues to advise the Department on decisions regarding tests and testing technology using Council of Regional Genetic Networks (CORN) guidelines as referenced in the American Academy of Pediatrics supplement "Serving the Family from Birth to the Medical Home, a report from the Newborn Screening Task Force Convened in Washington DC, May 10-11, 1999" published in August 2000.

Nebraska Human Genetics Technology Commission

The Nebraska Human Genetics Technology Commission was authorized during 1997/1998 legislative session and was active until December 2001. The Commission's central purpose was to encourage uses of human genetic technologies that contribute to the improvement of the human condition while assuring the protection of fundamental human rights. Guiding Principles were established by the Commission and were designed to serve as the governing context and standard of reference for all present and future policies, practices, laws, regulations and educational initiatives related to human genetic technology in the State of Nebraska. Implementation of this proposed State Plan would address one of the recommendations of the Commission which was to: "provide support for the Nebraska Health and Human Services System to establish a state advisory committee on clinical genetic services to address questions and concerns about access, cost and quality". Guiding principles on Human Genetic Technologies from the Commission Report have been used as the standard of reference for all recommendations set forth in the state genetics plan (Appendix G).

University of Nebraska Medical Center's Munroe-Meyer Institute for Genetics and Rehabilitation

The mission of the Munroe-Meyer Institute (MMI), the Nebraska University Affiliated Program, is to improve the quality of life for persons with disabilities and for their families. The Institute's services, interdisciplinary education, research, technical assistance, and outreach programs have evolved with the support and guidance of consumers. The services aim to promote independence, inclusion, and productivity of persons with disabilities in order to empower those individuals to exert greater control over their everyday lives. The Institute provides interdisciplinary family-centered services and supports for consumers and all individuals with disabilities. These services include the development of new and innovative ways to promote inclusion of the individual in the community.

MMI is administered by a director and a deputy director and is divided into 14 disciplines: developmental medicine, education and child development, genetics, human genetics laboratories, molecular genetics, nursing, nutrition, occupational therapy, orthotics, physical therapy, psychology, recreational therapy, social work, and speech-language pathology. The Media Center, Patient Information Office and the Business Office provide support and assistance to staff and students. Approximately 180 professional and support personnel operate the Institute.

A direct funding and sub-grantee relationship exists between the State's major genetics provider (UNMC/MMI) and the HHS Division of Family Health (MCH Title V). The Project Director and Director of the Center for Human Genetics at MMI, Dr. G. Bradley Schaefer, has worked collaboratively for over six years with the Principal Investigator and Newborn Screening and Genetics Program Manager, Julie Miller. Their efforts have included such issues as newborn hearing screening, folic acid education, promotion efforts for birth defects prevention, promotion of improved metabolic and hemoglobinopathy screening systems, and genetic education for Health and Human Services professionals and paraprofessionals via statewide Family Health conferences.

Eight of the members of the current Newborn Screening Technical Advisory Committee are from UNMC representing the genetics clinic and subspecialties of pediatric metabolism, pediatric endocrinology, pediatric hematology, pediatric metabolic dietetics, and laboratory services. In addition, the HHSS has support via contract for nearly 15 years, funding for metabolic formula and dietary consultation with the Metabolic Clinic at UNMC. The Newborn Screening Program has worked closely for nearly 10 years with the sub-specialty clinics to facilitate follow-up/retrieval, diagnosis and treatment. The UNMC/MMI collaborates extensively with Children's Hospital, which has the largest NICU in Nebraska, through clinics provided on myelomeningocele (neural tube defects) and cerebral palsy. UNMC health care professionals also with Creighton University/Boys Town National Research Hospital through clinics provided on craniofacial anomalies and hearing loss.

Leadership Education in Neurodevelopmental and Related Disabilities (LEND) Project

A major portion of the training program at MMI is funded through the Maternal and Child Health Bureau of Health Resources and Services Administration, U.S. Department of Health and Human Services. The MCHB Interdisciplinary Leadership Education in Neurodevelopmental and Related Disabilities (LEND) Project provides funding for long-term training for health professionals at the advanced graduate and post-graduate levels,

with a special focus on family-centered, community-based, culturally competent care. The specific purpose of the LEND Project is to improve the health status of infants, children, and adolescents with, or at risk for, neurodevelopmental and related disabilities, including mental retardation, neurological disorders, and multiple disabilities, and their families. This is accomplished through training of professional personnel for leadership roles in provision of health care, and through provision of continuing education, technical assistance and consultation.

Project LEARN, Leadership Education in Advancements in Research in Neuroscience

Project LEARN (Leadership Education in Advancements in Research in Neuroscience) is a grant promoting healthy children through the development of innovation strategies. Strategies include educating families, maternal and child health professionals, community providers, policy-makers and students about recent advances in neuroscience. A natural link between Project LEARN and this State Plan exists particularly in regard to developing infrastructure for access to quality services via statewide education on neurodevelopmental issues for children with special health care needs.

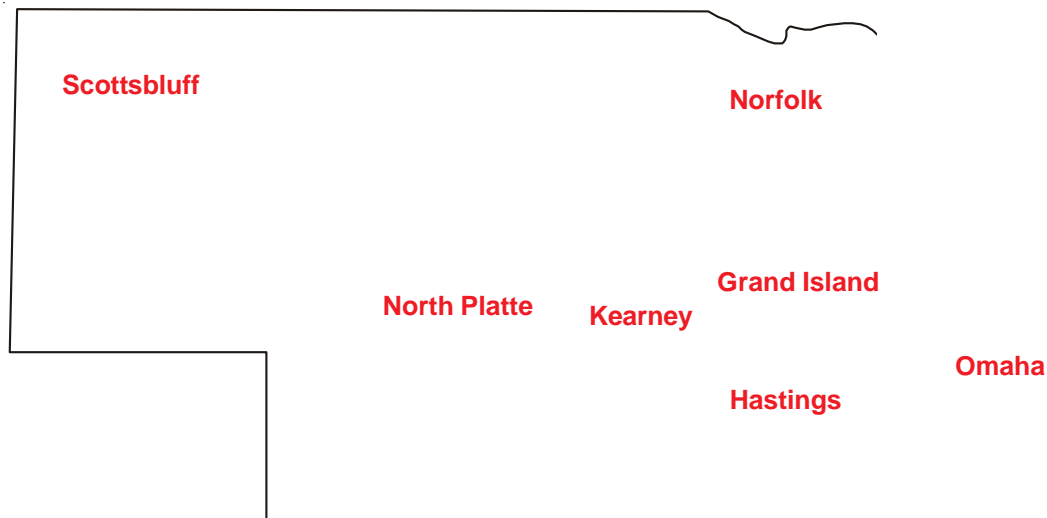
Tele-Health Project for Children with Special Health care Needs in Nebraska

Tele-Health Project for Children with Special Health Care Needs in Nebraska is a joint venture between MMI, HHSS, and the Nebraska Department of Education. Through this project a comprehensive link is being developed between the experts at MMI and local service providers for children with special health care needs throughout Nebraska. The Tele-Health Project provides another link to this State Plan by addressing the infrastructure need to improve access to specialized genetic services to remote and rural areas across Nebraska.

Support for Genetics Clinics Coordinated by UNMC/MMI

The genetic outreach clinics of MMI attempt to ensure access to underserved populations through quarterly outreach genetics/neurodevelopmental clinics and participation in four of the CSHCN cerebral palsy clinics at four sites; 16 multi-disciplinary clinics on regularly scheduled intervals; and, outreach diabetes clinics at three sites 3-7 times per year, and at two sites monthly. In Fiscal Year 2001 (7/1/01-6/30/02), these clinics reached 287 children with special health care needs and genetic disorders, and 857 pediatric patients with diabetes. The schedule represents a significant reduction in genetic outreach services, however, due to funding cuts experienced in 2000. With the outlook for increased funding being highly unlikely, it is critical that there be alternatives for reaching underserved populations by building infrastructure (e.g. telemedicine, expanded training of community based medical and health care professionals).

The following is a map of some of the locations where genetic outreach services have been provided. Due to major funding cuts in the program, Scottsbluff and Kearney will each host four clinics for the 2002 Fiscal Year.



Office of Minority Health and Human Services

Intra-program relationships between the Newborn Screening Program and the Office of Minority Health and Human Services in HHSS have been project specific. Collaboration has included facilitating development of special hemoglobinopathy screening task forces and identifying resources for translation of patient education materials into Spanish.

Birth Defects Sub-Committee

In 1999, the Governor of Nebraska, Mike Johanns, appointed an Infant Mortality Blue Ribbon Panel to address Nebraska's stagnant infant mortality rate, and increasing neonatal mortality rate. The Panel members represented a wide range of expertise from all parts of the state: neonatologists, epidemiologists, obstetricians, nurses, children's advocates, and consumers, as well as other individuals with expert knowledge. The group, acknowledging that infant mortality was the result of multi-faceted problems, chose to break into sub-committees to collect and analyze data, and develop recommendations. The Birth Defects Sub-Committee, active until 2000, was a group of persons interested in promoting the quality of the Birth Defects Registry, resources, and ability to utilize the data from the registry for active surveillance. This sub-committee made recommendations that have been incorporated into the State Plan (see Appendix G).

March of Dimes

Personnel from the NNSP and MMI have worked closely with the March of Dimes (MOD) by serving on the Nebraska Chapter's Program Services Committee for a number of years. This committee addresses issues affecting healthy birth outcomes. These three entities have collaborated closely to lead Nebraska's Neural Tube Defect Prevention Coalition, and to develop campaign strategies. The MOD also supported policy change in Nebraska in 2002 to expand the mandatory newborn screening panel.

Medically Handicapped Children's Program (MHCP – Nebraska's CSHCN)

The MHCP fills service gaps for infants and children needing diagnostic, medical treatment planning and medical treatment services through funding for genetics and CSHCN clinics across the state. Multidisciplinary teams, which include a MHCP service coordinator, are located in several communities. Since appropriate specialists and sub-specialists are not always available in the local communities, personnel from UNMC's MMI are contracted to complete the multidisciplinary team. Other teams made up of UNMC professionals, private specialists, and sub-specialists, travel to rural communities to conduct specialty care clinics. In 2003, the CSHCN program will sponsor over 50 Specialty Clinics for Children and Youth at five sites, 30 Craniofacial clinics at three sites, and 6 Midline Neurological defects clinics at one site.

Nebraska Teratogen Information Service

The Nebraska Teratogen Information Service provides information on hazardous exposures in pregnant and breast-feeding women. Hazardous exposures may include: prescribed and over-the-counter medications, herbal products, alcohol and street drugs, vaccines, infections, and chemical agents in the workplace or at home. Health care providers can request this information through the available hotline, and clinical consultations can be arranged for patients. This service is valuable in the prevention and awareness of birth defects caused by environmental exposures.

Early Development Network

The Early Development Network provides services and supports that are designed based on the needs of children birth to age three and their families with the belief that parents know what is best for their families. These services are designed to act on what families think is important for their child and family. The Network is staff by friendly, supportive people who listen to and respect families. The network also can connect families with other families who have had similar experiences.

The goal of the Early Development Network is to provide coordinated services for Nebraska families as conveniently as possible. The program helps families to understand their child's disability and provides assistance in dealing with situations that interfere with the child's development. Families may choose a particular service or all services. Families also may refuse services at any time. The program is co-administered by the Nebraska Department of Education and Health and Human Services.

As is obvious from the descriptions above, Nebraska has the personnel, capability, and willingness to reach out to under-served populations but this effort is hindered by decreasing funding and political support and the lack of an integrated database system for communicating needs and services.

Needs Assessment

The NBSGAC work group leaders and staff produced a prioritized list of needs for the Newborn Screening and Genetic Services programs/infrastructure by consolidating the input provided from the entire committee. As a group, the individual needs identified by all committee members were merged under nine main headings, listed here in no order of prioritization:

- ☐ Parent Education
- ☐ Physician Education
- ☐ Coordinated Information Management System
- ☐ Legislation
- ☐ HIPAA/Informed Consent
- ☐ Medical Home
- ☐ Transitioning
- ☐ Policies/Procedures
- ☐ Expanded Services

Within each of these headings, the needs were then prioritized and categorized sequentially according to how soon they might be addressed. The designation of “*short-term*” need was given to any need that might be met within two years, a “*medium-term*” need for less than five years, and “*long-term*” need for those needs possibly requiring five or more years. Some needs were also designated as requiring *continuous* improvement.

PARENT EDUCATION

The following needs were primarily identified based on the responses to the parent surveys (Appendices C and D), and the personal experiences of consumers of the CSHCN system and health care providers.

Short-term needs:

- ☐ Parent and patient education about the NNSP, mandatory vs. supplemental screening, what a “positive screen” result means, and for consent to genetic testing.
- ☐ Sufficient translation services for NBS and genetic services materials, educational efforts, and appointments for evaluation and follow-up.
- ☐ Improved communication between health care providers and parents regarding the NNSP and the possible results of the screening (negative, presumptive positive, positive).
 - A lack of communication by the primary care physician to the family when there is a confirmed positive result following the NBS, about the condition and future prognosis and care needs of the diagnosed infant has been expressed.
- ☐ Sufficient amount of information provided to parents whose infant has a confirmed positive result following a positive screen from the NNSP.
- ☐ Create a designated Emergency Information Form that will be available to parents.
 - The form should provide details on the child’s medical needs and history so that everyone involved in the child’s care has access to the information.

Long-term needs:

- ☐ An efficient method of connecting parents of newly diagnosed infants or CSHCN for emotional support.
- ☐ Financial support for parent-to-parent groups.

PHYSICIAN EDUCATION

The following needs were primarily identified based on the responses to the physician and parent surveys (Appendices B, C, and D), and the personal experiences of consumers of the CSHCN system and health care providers.

Short-term and continuous needs:

- ☐ Health care provider education about all aspects of the NNSP, emphasizing the need to discuss this information with their patients.
- ☐ Update the information in the NE NBS Practitioner's Manual and NNSP web site and redistribute the Manual and web site address.
 - The information provided in the Manual and on the web site requires regular review and revision.
 - The NNSP needs to establish a process that ensures this review.
- ☐ Awareness by primary care physicians of the national guidelines for CSHCN and the use of tandem mass spectrometry (MS/MS) in NBS.
- ☐ Education of health care providers and families/consumers about medium chain acyl co-A dehydrogenase deficiency (MCAD), the method of screening for MCAD using tandem mass spectrometry (MS/MS), and any other changes in the NBS program.
- ☐ Appropriate amount of information provided to physicians who have infants in their care that have a confirmed positive diagnosis following a positive screen through the NBS Program
- ☐ Continuous communication between health care providers and the NNSP about NBS results and diagnostic testing.
- ☐ A comprehensive resource directory complete with a glossary of terms, appropriate web sites, a listing of state and local services, and available funding sources for services.
 - The resource directory will require regular review and revision.

Medium-term and continuous need:

- ☐ Create follow-up testing guidelines and treatment protocols to be used by health care providers for supplemental screening results beyond those for MCAD, in other words, for those disorders not mandated for screening in Nebraska.
 - There is currently a lack of national guidelines to follow.

COORDINATED INFORMATION MANAGEMENT SYSTEM

The needs identified for a coordinated information management system were based upon the review of currently available databases and the personal experience of those utilizing the existing databases. The over-riding need in this category is to be able to make better use of the data collected in order to better serve families.

Short-term need:

- ☐ Update the NNSP database, to increase its capacity and to meet mandated requirements.

Medium-term need:

- ❑ Reduce the redundancy of data collection by coordinating the NBS database in real time with the birth registry and other databases. Consider linking the information that is currently entered into separate databases.

Medium to Long term need:

- ❑ Increase the ability of current databases to capture case management or long-term follow-up information, multiple diagnoses, and medical home/primary care physician information.

Long-term needs:

- ❑ Legislative and administrative funding and support for database linkage and updating databases.
- ❑ Ability to enter data for Tracking Infant Progress (TIPS, a Neonatal Intensive Care Unit follow-up program) at all locations (hospitals, clinics, etc).
- ❑ An efficient method of collection and dissemination of information on CSHCN, including the communication of all NBS results to health care providers.
 - A need to integrate, or link, all applications that capture information concerning CSHCN, including programs such as Medicaid, Medically Handicapped Children's Program (MHCP), Birth Defects Registry, Nebraska Newborn Hearing Screening Program, Early Intervention, and the NNSP, and is able to ensure quality assurance.
- ❑ A method of centralized long-term follow-up of infants, children, adolescents, and adults identified with disorders for which the NNSP screen.

LEGISLATION

The following needs were identified after reviewing the current Nebraska statutes and Federal Laws that apply to NBS and CSHCN services.

Short-term and continuous need:

- ❑ Establish an adequate sustainable funding structure for the NNSP.

Short-term need:

- ❑ Determine if HHSS has authority over linked or integrated data.
 - If the HHSS has insufficient authority, then there is a need for flexible legislation to allow sharing of public health data between public health programs (medium-term need).

Medium-term and continuous need:

- ❑ Statutorily recognize the Newborn Screening Technical Advisory Committee (NBSTAC) and authorize their ability to advise the NNSP.

Medium-term needs:

- ❑ Clear guidance and authority from the state legislature for state supported activities in the area of newborn screening for long-term follow-up data management, reporting and analysis.
- ❑ Revise, or adopt, legislation for integrating and coordinating CSHCN services.

Long-term needs:

- ❑ Resources for interagency sharing and services.
- ❑ Ability to determine if the funding streams for existing programs with databases allow for the merging and sharing of their data.
- ❑ Federal standards for MS/MS screening regarding what disorders should be screened, what levels of metabolites to use as cut-offs for screening, follow-up treatment for each screenable disorder, etc.

- ❑ Clear guidance and authority from the state legislature for state supported activities in the area of program effectiveness.

HIPAA/INFORMED CONSENT

These needs were identified following review of the federal HIPAA legislation and the most current federal health privacy regulations. In addition, the Nebraska statutes addressing “informed consent” were reviewed.

Short-term needs:

- ❑ Ensure that current data systems meet HIPAA regulations.
 - This includes the need to define reasonable roles and policies given our specific needs and personnel to conform with the “minimum necessary” portion of HIPAA, and to develop a “Notice of Private Practices” that outlines how information is collected, maintained, used, and re-released.
- ❑ Develop strict access definitions and protocols for viewing medical and other service program records.
 - Education needs to be provided to healthcare providers with regard to the use and implementation of these protocols.
- ❑ Develop specific authorization/informed consent for collection, maintenance, use, and release of genetic information.

Medium-term need:

- ❑ Resolution of the conflicting aspects of informed consent regarding NBS for metabolic diseases that are addressed in LB 432 affection Nebraska statutes §71-519 thru 71-524, 44-2816, and 44-2822.
 - LB 432 mandates informed consent for predictive and pre-symptomatic genetic testing, whereas the NE statutes mandate NBS for certain metabolic diseases without the need for informed consent.
 - LB 119, which was introduced in the 2003 Nebraska Unicameral session, would resolve these conflicts.

MEDICAL HOME

These needs were identified based on the experience of service program consumers and medical home providers, the abilities of current databases, and review of the American Academy of Pediatrics Newborn Screening Task Force Report “Serving the Family from Birth to the Medical Home”

Short-term need:

- ❑ Involvement of physicians in all communication regarding each CSHCN in their care.

Short-term and continuous needs:

- ❑ Education of physicians about their responsibilities as the medical home provider for CSHCN.
- ❑ Increased parental knowledge about the NNSP, in general, and more specifically, about the meaning of presumptive positive results.

Long-term needs:

- ❑ Ability of current database to reflect the CSHCN medical home and doctor information.
- ❑ Consistent communication with physicians about the results of NBS for all infants in their care, not only those with a presumptive positive screen.

TRANSITIONING

These needs were identified based upon review of the currently available transitioning services and agencies, as well as the experience of consumers utilizing those services.

Long-term needs:

- ☐ Efficient and sufficient transitioning process, including education of health care providers, for transferring the care of a CSHCN from pediatric specialists to adult health care providers.
- ☐ Increased number of specialists/physicians available to care for transitioning patients and adults with special health care needs in both urban and rural areas.
- ☐ Funding and insurance reimbursement/coverage for the care of transitioning adults with special health care needs.

POLICIES/PROCEDURES

The following needs were identified based on review of the current policies and procedure for applicable programs.

Short-term needs:

- ☐ Develop a protocol for communicating abnormal lab results for MCAD and the actions to be taken for confirmational testing or immediate dietary changes.
- ☐ Develop an evaluation tool utilizing objective measures for adding and/or deleting diseases to be screened for in the mandatory NBS panel. Currently, both the Newborn Screening Technical Advisory Committee (NBSTAC) and the American College of Medical Genetics (ACMG) have developed, but not tested, evaluation tools.
- ☐ Increase the membership of the current NBSTAC, as it lacks some of the recommended representation as outlined in "Serving the Family from Birth to the Medical Home" by HRSA, AAP, AHRQ, AMCHP, APHL, ASTHO, CDC, Genetic Alliance, and the NIH.
- ☐ Administrative and personnel support of interagency data sharing and services relative to CSHCN.

Medium-term needs:

- ☐ Identification of term infants with special health care needs and the corresponding connection to services in early infancy/childhood.
 - n Pre-term infants and those who have been in the NICU can be identified and connected with services through the TIPS program.
- ☐ Service coordination policies within HHS.
 - Need for a central repository to help families navigate through complex payer systems. Families need to know if services are going to be paid for by their insurances, or if they don't have insurance coverage, if they will get financial help through the CSHCN agencies.
 - The need for a listing of what financial resources are available to help families to meet the costs of services (lab, follow-up, treatment, etc).

Long-term needs:

- ☐ Quality assurance to assess service quality and efficiency and cost-effectiveness. These efforts are hampered by the lack of published information on the cost-effectiveness and treatment effectiveness for those disorders available on the expanded NBS, not those mandated for screening in Nebraska.
- ☐ A coordinated, non-fragmented system of coverage of medical care.

- ❑ Long-term, sustainable funding for the NNSP and services for CSHCN.

EXPANDED SERVICES

Many of the following needs were identified after comparing the current services available in Nebraska to those recommended in the American Academy of Pediatrics Newborn Screening Task Force Report “Serving the Family from Birth to the Medical Home”.

Short-term and continuous needs:

- ❑ A dedicated full-time State Genetics Coordinator position, a permanent broad-based Genetics Advisory Committee, a full-time Medical Advisor for the NNSP, and a state-supported Educator position to implement the needs identified by this document.
- ❑ Sufficient personnel and resources for expanding the NNSP beyond those diseases currently mandated.
- ❑ Increased availability of respite care, adequate mental health care and pediatric providers for families of CSHCN.
- ❑ Increased access to oral and behavioral health care providers for CSHCN.
 - Previous needs assessments have determined that there are not enough dentists trained to care for CSHCN, not enough dentists accepting Medicaid patients or not signed up with Medicaid to accept patients.
- ❑ Specialists with training in the care of CSHCN once they reach adulthood.
- ❑ Funding for services for certain patient needs (such as testing phenylalanine levels in pregnant women with PKU to prevent mental retardation in the unborn children).

Medium-term needs:

- ❑ Involvement of genetic counselors with patients who have metabolic disorders, other single gene disorders, adverse reproductive outcomes, congenital anomalies, and positive family history including common disorders such as cancer and diabetes. Additionally, need to involve genetic counselors with all patients who are diagnosed through the NBS, except those with congenital primary hypothyroidism (all others are monogenic, or single gene, disorders). Genetic counseling will assist the family in understanding the cause, inheritance, and recurrence risks for the disorder.
- ❑ Resources for personnel, increased operating costs, and treatment of underserved/poorly-served populations following screening of additional disorders, beyond MCAD, as the current system is currently at maximum capacity.
- ❑ Increased access to metabolic clinics to care for patients on an on-going basis.

Long-term needs:

- ❑ Additional transitioning, adult services and treatment programs for adults with special health care needs.
- ❑ A fee structure which keeps pace with the cost of implementing, maintaining, improving, and administering the NBS system.
- ❑ Funding and adequate staffing to design, develop, and maintain the desirable database system.
 - The desirable database system is a system that links current databases, those used for surveillance and tracking, which makes it useful and accessible to physicians and pediatric specialists.

- ☐ Increased and accessible funding to support activities for CSHCN.
- ☐ Competent, financially rewarded staff for Developmental Disabilities (DD) services, especially the need for incentives to keep trained, knowledgeable staff. A loyal staff will ensure consistency in patient care, including those with specialized diets.
- ☐ Increased access to services available to those with mental retardation or developmental disabilities.
- ☐ A centralized state agency or location for collection and dissemination of information on genetic disorders and the coordination of State of Nebraska activities to respond appropriately to these disorders.

Family Story #3

Les was born July 11, 1978 in Norfolk, Nebraska. He appeared to be a healthy baby from the start, with the exception of not eating well. We left the hospital when he was four days old. All of the required newborn screenings were done before we left. At Les' three-week check, our local doctor did the diaper check for PKU and determined that it was negative. A couple of days later, we received a call from the local lab responsible for the lab work done at the hospital on Les. They wanted us to return to the lab for another PKU test. I stopped by the doctor's office on the way to the lab and inquired about the reason for another test. The doctor said that the second test was not necessary. Then I went to the lab to discuss why they wanted another test. I was informed at the lab that the screening done on Les when he was three days old looked suspicious for PKU and they wanted to screen Les again. We allowed them re-screen Les and informed the doctor that we did. Again the doctor said it was unnecessary because "PKU was rare and only ½ a baby per year was diagnosed with PKU in Nebraska." Two days later we received a call with the lab report that the PKU test again looked suspicious and we needed to go to University Medical Center in Omaha with Les.

On the Saturday, we traveled to Omaha to the Metabolic Disorders Clinic and University Medical Center and met with Dr. Hobart Wiltse and Mary Balluf. Dr. Wiltse did additional PKU screenings and even before the results of the tests were compiled, put Les on the formula for PKU and assumed that he had PKU based on the results of the Norfolk lab. In the next few days we learned the Les, indeed, did have PKU. Mary Balluf was the nutritionist that helped us adjust to the PKU treatment for Les. She also helped us adjust to the fact that we did not have the perfect baby. Mary was a temporary nutritionist while Betty Romano was on maternity leave. She was the mainstay of our life for the first six weeks of treatment for Les. Betty Romano became a wonderful support person for us dealing with all the aspects of PKU and all the issues a new parent has. Betty Romano and Dr. Wiltse worked with us for over twenty years. Betty is now deceased and Dr. Wiltse has retired, but both were vital in Les' health and development. The people at the UN Medical Center are still monitoring Les and Jill Skrabal is the nutritionist now.

Without the law requiring genetic screening for PKU, Les would have become retarded. The local doctors were not informed about PKU and/or weren't knowledgeable about the effects. There is a boy just older than Les that was not screened for PKU at our local hospital, and he also has PKU. The results are severe retardation and legal actions found the hospital and the local doctor negligent.

Without the competence of the local lab technicians, Les would also have been retarded. They are the ones who noticed the suspicious test results and wanted a follow up. They went ahead with the screenings without the support of Les' doctor.

Without the competence of the Metabolic Disorders staff of the UN Medical Center, Les would not be the brilliant and successful adult that he is. Les recently graduated with honors, from Baylor University with a Master's degree in Business Administration. Dr. Wiltse, Betty Romano, and the entire staff and the Medical Center are to be thanked for that.

The story continues with the birth of our second son on July 12, 1980. Lee was born at a different hospital with a different group of doctors in control. With the support of all medical personnel involved, Lee was screened for PKU several times before we accepted the fact that he DID NOT have PKU. The people at the University Medical Center again supported us with all the information and testing that was necessary.

This past September, we again needed the Newborn Screening Program when our first grandson was born. Lee's son, Andrew, does not have PKU, but was tested and retested for our peace of mind. When Andrew was born, Lee signed to have all available genetic screenings done. I would think that if there were a test that can screen for 60 plus disorders, the State of Nebraska should require that it be done, instead of making it optional.

We have been very pleased with the State of Nebraska and their Newborn Screening and Genetics staff.

Recommended Actions for the State

The needs identified in the assessment process became the basis for following recommended actions. The actions provide strategic planning for the current services, as well as propose development of necessary service infrastructure. The goal of this State Plan is that children with special health care needs with genetic disorders will receive family focused, community-based, culturally competent, comprehensive genetic health care. This includes outreach, intervention, education, and integration of the public health and private sector arenas for as outlined in *Healthy People 2010* Goals and Aims (Appendix I).

The NBSGAC determined that six core areas of action could address the nine previously detailed areas of need:

- ☐ Education of both parents and healthcare providers/physicians
- ☐ Financial support to implement these actions
- ☐ Personnel for staffing these services
- ☐ Legislative action, policies and procedures to authorize the actions,
- ☐ A coordinated information management system (database) for effective communication
- ☐ Expanded services, especially those in transitioning, long-term care, and medical home implementation

The following are the specific recommended actions required to meet the identified needs.

A. EDUCATION

1. Review and update all professional and patient education materials using a broad range of media to reflect current advances.
 - a. Create a brochure for parents about the NNSP and the cause of presumptive positive or positive NBS results. Make the brochure available in hospitals, physician's offices, and on-line.
 - b. Reinstate preparing and distributing NBS newsletters.
 - c. Use and develop, as needed, specific fact sheets for parents, physicians, and other health care workers, of frequently asked questions (FAQs) for each disorder screened, to send to the primary care physician when they, or a specialist, are notified by the NNSP of a positive screening result. Include information about each screened condition, the screening methods employed, follow-up testing requirements for presumptive positive screens, appropriate referral and treatment, and prognosis.
 - d. Prepare an educational newsletter for health care providers and patients/families about Medium Chain Acyl Co-A Dehydrogenase deficiency (MCAD), the method of screening for MCAD using tandem mass spectrometry (MS/MS), plus any other changes in the NNSP. Consider distributing this information to health care providers offering obstetrical care to supply to parents when they are asked to consent or dissent to supplemental NBS.
 - e. Prepare and distribute a packet for parents of infants with a confirmed positive NBS that includes the following information specific to the confirmed diagnosis:
 - A description of the specific diagnosis – *This is already available for each diagnosis.*

- Treatment information specific to the diagnosis
 - Information on what will need to be done to provide for child, such as a brief description of the diet or medication
 - Contacts for support and referral information (agencies, specialists, etc) – *Currently available for hemoglobinopathies only.*
 - Reliable web sites for further information regarding the diagnosis
 - Questions and answers section
 - Consent form so their name can be given to another support family, if desired.
- f. Modify the current protocol of communicating with physicians regarding infants in their care that have a confirmed positive diagnosis by preparing and distributing a packet that includes the following information specific to the confirmed diagnosis:
- A copy of the test results – *Already part of the current protocol*
 - A description of the disease
 - Further tests required to confirm or rule-out a diagnosis – *Already part of the current protocol*
 - Contacts for support agencies and specialists – *Already part of the current protocol*
 - Reliable web sites
 - A list of questions parents may ask and the corresponding answers
 - Referrals to provide parents
- g. Develop, adapt, or acquire an Emergency Information Form (EIF) to the families of newborns diagnosed with a disorder by NBS or genetic services so that it is available for schools and daycare providers, and will ensure quick access to pertinent health care information. The form should provide details regarding the child's medical needs and history. In addition, the Medical Home Providers should stress to the families the need to provide this information to everyone involved in the child's care.
- h. Update and distribute the State Practitioner's Manual for NBS and NNSP website to include MS/MS screening information. Inform providers that, currently, there are no published guidelines for the other disorders detected by MS/MS (supplemental screen), and there exists inconsistent professional agreement on best practice.
- i. Prepare and distribute a brochure to increase the educational efforts of patients about the need to consent to genetic testing (beyond that of newborn screening).
2. Develop a working relationship with those providing obstetrical care in Nebraska.
3. Develop and promote in-service training for those providing obstetrical care on educating their parents-to-be about the NNSP, including the differences between mandatory and supplemental screening. Emphasize the need to discuss NBS with their parents-to-be.
4. Collaborate with those providing obstetrical care to provide information and suggestions on how to implement written materials (developed by the program) to mothers during the third trimester of their prenatal care.
5. Ensure the parent's receipt of the NBS results via the infant's primary care physician by establishing a dependable method of communication.

6. Develop and distribute reminders for primary care physicians advising them of the national guidelines for CSHCN, including the coordination of CSHCN's services as part of being the medical home provider.
7. Revise the NNSP web site and advertise the program's phone number and business hours so parents and physicians can readily access and review NBS information. Include links to the NE Parent NBS Guide and to resourceful, reliable websites such as those for the Massachusetts NBS Program, the National Newborn Screening and Genetics Resource Center (NNSGRC), or other worthy sites.
8. Create the initial follow-up guidelines/protocols for supplemental screening results beyond those for MCAD (for those disorders not mandated for screening in Nebraska), as there currently is a lack of national guidelines to follow.
9. Create a comprehensive resource directory complete with a glossary of terms, listing of state and local services, and available funding sources for services to be distributed to physicians and service providers.
10. Create a listing of what financial resources are available to help families to meet the costs of services (lab, follow-up, treatment, etc).

B. FUNDING

1. Obtain funding to support the activities of, and ensure adequate staffing for design, development, and maintenance of a desirable database system, either by linking and updating the current databases or creation/implementation of a new ideal system.
2. Obtain additional resources for personnel, increased operating costs, and funds for treatment for underserved/poorly-served populations (geographic, ethnic, and fiscal) for screening of additional disorders, beyond MCAD. The current staffing and funds are at maximum capacity and increasing the number of disorders screened will necessitate an increase in the NBS infrastructure.
3. Establish funding to support activities for CSHCN, such as:
 - a. Addressing the very high turnover rate for developmental disability services, by hiring competent staff that are financially rewarded.
 - b. Expanding and improving funding for services beyond screening, especially in support of outreach clinics.
 - c. Establishing a database for supportive parents of CSHCN who are willing to talk to other parents, and which extends to families moving into and out of the area.
 - d. Increasing the financial support for parent-to-parent groups for meetings, mailings, food, supplies, etc.
4. Create a method of long-term, sustainable funding for these services.
5. Allocation issues require addressing rather than an overall need for funds.

C. PERSONNEL

1. Recruit and train specialists/physicians to care for CSHCN and transitioning adults with special health care needs.
 - a. Fund training initiatives in post-graduate health care education as they relate to providing the medical home for individuals with special health care needs.
 - b. Hire additional personnel and encourage students to train in the respite care, mental health and pediatric fields to address the lack of availability of respite care and adequate mental health and pediatric providers in Nebraska.

2. To ensure adequate representation on the NBSTAC, recruit representatives from a Pediatrics/Family Practice medical home, an additional Family Practitioner, parents and/or general public, an insurance industry representative and a medical ethicist, as outlined in "Serving the Family from Birth to the Medical Home" by HRSA, AAP, AHRQ, AMCHP, APHL, ASTHO, CDC, Genetic Alliance, NIH.
3. Establish a dedicated full-time State Genetics Coordinator position.
4. Establish a permanent broad-based Genetics Advisory Committee.
5. Establish a state-supported Educator position.
6. Accommodate the expansion of NBS (beyond the diseases currently mandated) with increased personnel and resources.

D. LEGISLATION

1. Establish an adequate long-term, sustainable funding structure for the NNSP to address the serious and persistent deficit issues. The fee structure should keep pace with the cost of implementing, maintaining, improving, and administering the NBS system and relieve dependence on grant funds.
2. Establish broad authority to share data for purposes of quality assurance and service provision.
 - a. Legislative review of current regulations/statutes and action to allow funding streams allow for merging and sharing of data (categorical programming) and determine if HHSS has authority over linked or integrated data. If there is not sufficient authority, then create flexible legislation to allow sharing of public health data between public health programs. Request clear guidance and authority from the state legislature for state supported activities in the area of newborn screening for long-term follow-up data management, reporting and analysis. There are many ongoing programs for the improvement of health related data collection and management. HHSS, CDC, and private vendors are involved in these projects. Specific guidance from the legislature would ensure newborn screening was a part of this process.
 - b. Revise, or adopt, legislation for integrating and coordinating CSHCN services.
3. Statutorily recognize the NBSTAC (Newborn Screening Technical Advisory Committee) and authorize them to advise the Department, in order to ensure continued support of this entity.
4. Work with interested state legislators and non-governmental groups to propose legislation that will allow the state to develop a comprehensive and unified system to support all aspects of NBS and CSHCN data. Earmark funds specifically for these programs. (Data Committee)
5. Await, then adopt, federal standards for MS/MS screening regarding what disorders should be screened, what levels of metabolites or algorithms to use as cut-offs for screening, and follow-up treatment for each screenable disorder, etc. (federal standards are said to be coming in the distant future).
6. Resolve the conflicting Nebraska legislation that address "informed consent." (LB 432, NE Statutes §71-519 to 71-524, 44-2816, and 44-2822). *As mentioned earlier, LB 119, introduced to the Unicameral in 2003, would resolve these conflicts.*

E. DATABASE

1. Update and modernize the current NBS database program and expand its capacity and reduce the redundancy of data collection by coordinating the NBS database in real time with the birth registry and other applicable databases. The coordination of data collection may be made by linking the information that is currently entered into separate databases.
2. Design, develop, and maintain an integrated computer application that:
 - a. Captures the necessary information on newborns
 - b. Integrates, or links, all applications that capture information concerning CSHCN, including programs such as Medicaid, Medically Handicapped Children's Program, Birth Defects Registry, Nebraska Newborn Hearing Screening Program, Early Intervention, and the NNSP.
 - c. Allows for NBS results to be readily available to providers for review to ensure that the infants in their care have been screened.
 - d. Involves physicians in all communication regarding each CSHCN.
 - e. Ensures appropriate evaluation, performance monitoring, and quality assurance (capability) activities from the initial screening tracking through diagnosis, treatment, follow-up, and services.
 - f. Captures follow-up information, multiple diagnoses, and CSHCN medical home or primary care physician. Currently, the database won't capture information past the time of diagnosis and intervention and CONNECT is only able to capture the primary diagnosis.
 - g. Creates a centralized location for collection and dissemination of information on CSHCN that is available to providers through modern communications.
3. Enable all TIPS (Tracking Infant Progress, a Neonatal Intensive Care Unit follow-up service program) data to be put into the same database for all locations, which will ease evaluation of outcome data.
4. Ensure that current data systems meet HIPAA regulations. This includes the need to define reasonable roles and policies given each program's specific needs and personnel to conform with the "minimum necessary" portion of HIPAA, and to develop a "Notice of Private Practices" that outlines how information is collected, maintained, used, and re-released.
 - a. Develop and enforce strict access definitions and protocols for viewing medical and other service's records.
 - b. Develop and require specific authorization/informed consent for collection, maintenance, use, and release of genetic information.
1. Request a commitment for administrative and personnel support of interagency data sharing and services relative to CSHCN.

F. SERVICES

1. Increase and improve translation services for NBS and genetic services materials, educational efforts, and appointments for evaluation and follow-up.
2. Create an organized transition process for transferring care from pediatric specialists to an adult health care provider.

3. Identify and connect term infants with special health care needs to services in early infancy/childhood by expanding the role of the TIPS program (currently, only pre-term infants can be identified and connected with services through the TIPS program).
4. Continuous research for more published information on cost-effectiveness and treatment effectiveness for those disorders available on the expanded NBS and not mandated for screening in Nebraska.
5. Improve and increase services for certain patient needs such as testing phenylalanine levels in pregnant women with PKU to prevent mental retardation in the unborn children.
6. Involve genetic counselors with patients who have metabolic disorders, other single gene disorders, adverse reproductive outcomes, congenital anomalies, and positive family history including common disorders such as cancer and diabetes. Additionally, need to involve genetic counselors with all patients who are diagnosed by the NBS, except those with congenital primary hypothyroidism (all others are monogenic, or single gene, disorders).
7. Address the waiting lists for services available to those with mental retardation and developmental disabilities by increasing transitioning, adult services and treatment programs for those with special health care needs.
8. Revise and implement an effective evaluation tool utilizing objective measures for adding and/or deleting diseases to be screened for in the mandatory NBS panel.
9. Increase access to metabolic clinics to care for CSHCN on an on-going basis.
10. Improve the access to pediatric sub-specialists through telemedicine, increased outreach clinics, etc.